CURRICULUM VITAE Karen E. Weck, M.D.

PERSONAL INFORMATION

Name: Karen Elizabeth Weck

Home Address:

Home Phone:

EDUCATION AND TRAINING

Research Fellow, Department of Pathology,

Washington University School of Medicine, St. Louis, Missouri

Chief Resident in Laboratory Medicine

Washington University School of Medicine, St. Louis, Missouri

Resident in Laboratory Medicine

Washington University School of Medicine, St. Louis, Missouri

Research Internship, Laboratory of Molecular Microbiology

National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Maryland

Duke University School of Medicine, Durham, NC

M.D. awarded May 8, 1988

Duke University, Durham, North Carolina

B.S. in Zoology, cum laude, 1984

Specialty certification

2003 Molecular Genetic Pathology (American Board of Pathology/ American

Board of Medical Genetics)

1993 Clinical Pathology (American Board of Pathology)

PROFESSIONAL EXPERIENCE

2010-present Clinical Professor of Pathology and Laboratory Medicine

Clinical Professor of Genetics (joint appointment)

Director of Molecular Genetics

Associate Director of Molecular Pathology

University of North Carolina School of Medicine, Chapel Hill, NC

2007-2013 Associate Director

UNC Institute of Pharmacogenomics and Individualized Therapy

2004-2010	Associate Professor of Pathology and Laboratory Medicine Director of Molecular Genetics Associate Director of Molecular Pathology University of North Carolina School of Medicine, Chapel Hill, NC
2008-2010	Associate Professor of Genetics (joint appointment) University of North Carolina School of Medicine
1999-2004	Assistant Professor of Pathology and Assistant Director Molecular Diagnostics, University of Pittsburgh Medical Center, Pittsburgh, PA
1995-1998	Research Instructor in Pathology, Washington University School of Medicine St. Louis, Missouri

HONORS AND AWARDS

Philip M. Blatt Award for Excellence in Resident Training in Clinical Pathology	2008
Young Investigator Award, Academy of Clinical Physicians and Scientists	1996
Clinician Investigator Award (K08), NIAID, NIH	1995
American Cancer Society Physician Research Training Award	1995
Young Investigator Award, Academy of Clinical Physicians and Scientists	1992
Young Investigator Award, Academy of Clinical Physicians and Scientists	1991
National Institute of Neurological Disorders and Stroke Post Doctoral Fellowship	1990
Early Identification Program Acceptance to Duke Medical School	1985

Invited Seminars and Presentations:

- 1. "Clinical Molecular Testing for Secondary Drug Resistance in Cancer," Next Generaton Dx Summit, Washington, DC, August 22, 2013.
- 2. "Genetic Testing in the Era of Personalized Medicine," North Carolina society of Pathologists Annual Meeting, Asheville, NC, April 5, 2013.
- "Laboratory Performance on Molecular Genetic Proficiency Testing," College of American Pathologists Workshop, American College of Medical Genetics Annual Meeting, Phoenix, AZ, March 19, 2013.
- "Clinical Genomic Testing in the Era of Personalized Medicine," Speck-tacular Gammaherpesvirus Research Symposium, Department of Microbiology and Immunology & Emory Vaccine Center, Emory University, February 16, 2013.
- 5. "Advances in Molecular Diagnostic Cancer Testing," Next Generation Diagnostics Summit, Washington, DC, August 20, 2012

- 6. "Application of Pharmacogenomic Technologies in the Clinical Laboratory," 2nd Latin American Pharmacogenomic and Personalized Medicine Congress, Rio deJaneiro, Brazil, June 28-29, 2012.
- 7. "Pharmacogenomic Testing to Direct Clinical Therapy at UNC," Gentris Corporation, Research Triangle Park, Morrisville, NC, December 15, 2011
- 8. "Genetic Testing Principles Applied to Case Studies: BRCA1 and BRCA2 Analysis & Cystic Fibrosis Mutation Analysis" Association for Molecular Pathology Outreach Course, Dallas, TX, November 16, 2011.
- 9. "Pharmacogenomic Testing to Individualize Cancer Therapy," Next Generation Diagnostics G2 Summit, Washington, DC, August 22, 2011.
- 10. "Anecdotes of Success in Personalized Medicine: Pharmacogenomics," Association for Pathology Chairs Annual Meeting, Monterey, California, July 13-15, 2011.
- 11. "Pharmacogenetic testing", Duke School of Medicine Department of Genetics, June 29, 2011
- 12. "CYP2D6 Genotyping to Guide Use of Tamoxifen in Breast Cancer," 21st International Congress of Clinical Chemistry and Laboratory Medicine, Berlin, Germany, May 15-19, 2011.
- 13. "Pharmacogenomics in the Clinical Laboratory," G2 Intelligence Conference on Molecular Diagnostics, Boston, MA, April 13-15, 2011
- 14. "Case Studies in Molecular Genetics: Cystic Fibrosis Mutation Analysis and BRCA1/2 Analysis", Association for Molecular Pathology Outreach Course, San Jose, CA, November 17, 2010.
- 15. "CYP2D6 Genotyping to Guide use of Tamoxifen in Breast Cancer", Symposium on Pharmacogenomics in Clinical Practice, American Association of Clinical Chemistry annual meeting, Anaheim, CA, July 29, 2010.
- 16. "The Multidisciplinary Approach to Personalized Medicine: Fitting together the pieces of the 'P-4' Puzzle (Predictive, Preventive, Personalized, and Participatory)". Invited panelist, Conference on Personalized Medicine in the 21st Century, RTI International and the North Carolina Biotechnology Center, Durham, NC, June 15, 2010.
- 17. "Pharmacogenetic testing for individualized therapy" American Association of Clinical Chemistry, Capital Section Annual Spring Meeting, Richmond, VA, May 13th, 2010
- 18. "Genetic Testing for Personalized Medicine *Are we there yet*?" North Carolina Medical Genetics Association Annual Meeting, Asheville, NC, April 16, 2010.
- 19. "Pharmacogenetic testing", Duke School of Medicine Department of Pathology, June 2009
- 20. "ABL1 Kinase Mutation Analysis", Invited speaker and panel participant, Association of Molecular Pathology Annual Meeting Workshop, Dallas, TX, Nov 2, 2008
- 21. "GeneticTesting Principles Applied to Case Studies: Cystic Fibrosis, BRCA1/2, and DNA Sequencing Analysis", College of American Pathologists Annual Meeting, San Diego, CA, September 30, 2008.
- 22. "Genetic Testing for Identity, Medical Diagnosis and Forensic Analysis", International Judicial Academy Conference in Mendoza, Argentina and Santiago Chile, May, 2008

- 23. "The Right To Health", Speaker and Panel Participant, International Judicial Academy conference in Buenos Aires, Argentina, May 2008
- 24. "Genetic Testing for Primary Ciliary Dyskinesia", Annual Update in Clinical and Laboratory Medicine, Park City, Utah, March 3-7, 2008
- 25. "Pharmacogenetic Testing for Warfarin Response", Annual Update in Clinical and Laboratory Medicine, Park City, Utah, March 3-7, 2008
- 26. "Mutation detection in the clinical laboratory: the new frontier of genomic medicine", UNC Department of Genetics Research Colloquium, February 13, 2008
- 27. "Implementation of a Program to Aid in Warfarin Dosing", Invited speaker and panel discussant, Association for Molecular Pathology Annual Meeting Workshop, Los Angeles, CA, November 10, 2007
- 28. "Genetic Testing Principles Applied to Case Studies: Cystic Fibrosis, BRCA1/2, and DNA Sequencing Analysis", College of American Pathologists Annual Meeting, Chicago, IL, October 2, 2007.
- 29. "Genetic Testing for Focal Segmental Glomerulosclerosis", 22nd Annual Meeting of the Glomerular Disease Collaborative Network, Chapel Hill, NC, May 19-20, 2007
- 30. "Individualized Therapy", Session Chair, 2nd Annual Chapel Hill Drug Conference: Pharmacogenomics, May 18, 2007
- 31. "Molecular Classification of Cancer: Practical Applications for the Surgical Pathologist", Organizer and Moderator, AMP companion meeting, USCAP Annual meeting, San Diego, CA, March 2007
- 32. "Development and Quality Control of Sequencing Assays," Workshop Moderator and Presentation of an Unusual Case, Association for Molecular Pathology Annual Meeting, Orlando, FL. November 2006
- 33. "New Applications of Mutation Detection in Diagnostic Medicine," UNC Department of Pathology and Lab Medicine Annual Research Symposium, Chapel Hill, NC, September, 2006
- 34. "An Interesting Fragile X Case," Genetic Puzzlers Workshop, Association for Molecular Pathology Annual Meeting, Scottsdale, AZ, November 2005
- 35. "Development and validation of real-time PCR assays for viral load monitoring: CMV and BK viruses," Workshop Moderator, Association for Molecular Pathology Annual Meeting, Los Angeles, CA, November 2004.
- 36. "Quantitative PCR for Diagnosis and Monitoring of BK Virus Post-transplant Nephropathy," Academy of Clinical Laboratory Physicians and Scientists Annual Meeting, Denver, CO, June 2004.
- 37. "Molecular Testing of Antibiotic Resistance," Workshop Moderator, Association for Molecular Pathology Annual Meeting, Orlando, FL, 2003.
- 38. "Macrophages are a Major Reservoir of γ HV68 Latency," 24th International Herpesvirus Workshop, 1999

- 39. "B cells are important for clearance of murine γHV68 latency *in vivo*," American Society for Virology 17th Annual Meeting, 1998
- 40. Workshop on Gammaherpesviruses, 22nd International Herpesvirus Workshop, 1997
- 41. Workshop on Gammaherpesviruses, 21st International Herpesvirus Workshop, 1996
- 42. Academy of Clinical Laboratory Physicians and Scientists Annual Meeting, 1996
- 43. XVIII Symposium of the International Association for Comparative Research on Leukemia and Related Diseases, Kyoto, Japan, 1995
- 44. Academy of Clinical Laboratory Physicians and Scientists Annual Meeting, 1992
- 45. Academy of Clinical Laboratory Physicians and Scientists Annual Meeting, 1991

Interviews

Genomics and Personalized Medicine in Pathology at the University of North Carolina, USCAP TV, March 2012.

Next Generation Sequencing in the Clinical Laboratory. CAP today, April 2011.

Experts split on need for greater FDA oversight of diagnostic tests. Elsevier Global Medical News, MD consult www.mdconsult.com. March 4, 2009.

PGx tests for warfarin dosing – how soon? CAP Today, January 2009, Feature story

Too fast or too slow on PGx testing? CAP Today, March 2008, cover story.

Pharmacogenomics enables more targeted treatment. Cancer Lines, UNC Lineberger Comprehensive Cancer Center Newsletter, Spring, 2007.

Are drug companies stalling the pharmacogenomic revolution? Diagnostic Testing and Technology Report. June 2004, cover story.

Welcoming resistance tests, old and new. CAP Today. May 2004, cover story.

TEACHING RECORD

Research Advisory Committees and Mentorships

2011-present	PhD Thesis Advisory committee, University Dept of Biomedical Engineering	, North Carolina State
2007-2009	Research Advisor/ Mentor for Holderness Distinguished Medical Scholar	MSIII, UNC School of Medicine
2001	Faculty mentor for Hepatology, University of Pittsburgh, to con investigating diversity in hepatitis C and cor orthoptic liver transplantation. ~25 contact h	relation with clinical outcome in

Lectures/ small group teaching

2011 "Pharmacogenomic Testing to Predict Response to Cancer Therapy," Genomics in Society course, UNC School of Nursing, April 8, 2011.

2010-2011	Course Director, "Current Applications of Molecular Pathology: Real time updates and case studies," Association of Molecular Pathology Outreach Course, November 17, 2010 San Jose, CA and November 16, 2011, Dallas, TX
2010-2011	"Pharmacogenomics in Clinical Practice," Fourth year Medical Student Basic Science Elective, UNC School of Medicine
2009-present	"Pharmacogenomic Testing in the Clinical Laboratory," DPET 832 Introduction to Applied Pharmacogenomics, UNC Eshelman School of Pharmacy Graduate course. 2 contact hours
2010	"Genetic Testing in the Era of Personalized Medicine, Genomics in Society Course, UNC Nursing and Undergraduate, October 19, 2010, 2 contact hours
2010-present	"Pharmacogenetics," Medical Genetics course for clinical genetic fellows and residents, UNC School of Medicine
2007-2009	"Genetic Testing Principles Applied to Case Studies: Cystic Fibrosis, BRCA1/2, and DNA Sequencing Analysis", Half Day Course, College of American Pathologists Annual Meeting.
2007-present	Lecture "Translating Genetics to Clinical Medicine," Translational Pathology and Laboratory Medicine Graduate Course, University of North Carolina. <i>2 contact hours</i>
2007-2011	Small Group Learning lab preceptor for 1st year Medical Student Course in Cell and Molecular Biology: Molecules to Cells, University of North Carolina School of Medicine. 6 contact hours, ~20 medical students.
2006-present	Lecture "Pharmacogenetics" for 2 nd year Medical Student Course in Reproductive Biology and Genetics, University of North Carolina School of Medicine 1 contact hour, ~150 students
2006-2010, 2013	Small Group Learning lab preceptor for 2 nd year Medical Student Course in Reproductive Biology and Genetics, University of North Carolina School of Medicine. <i>5 contact hours</i> , ~20 <i>medical students</i> .
2005-present	Director of Molecular Case Conference (monthly conference) presented by fellows in molecular pathology, University of North Carolina School of Medicine
2005-present	Attending supervision and teaching of clinical fellows in Molecular Genetic Pathology and Clinical Molecular Genetics, University of North Carolina
2005-present	Molecular Diagnostics/Cytogenetics course for Pathology residents and fellows, University of North Carolina School of Medicine. Lectures on "Pharmacogenetics", "Melting Curve Analysis," "Fragile X and other Triplet Repeat Disorders," "RT-PCR for Promyelocytic Leukemia," "Diagnostic testing for Hepatitis C virus," "Gleevec resistance testing"; Workshops on "PCR, RT-PCR, real-time PCR"; "Interpreting Sequencing assays"; "Melting Curve analysis and Mutation scanning"; "Interpreting Cystic Fibrosis testing"; "Interpreting Fragile X testing"
2006-2007	Lecture "Dominant Negative Mutations" to Genetics Residents and Fellows, University of North Carolina
2006	Lecture "Molecular Genetics" to graduate students, Methods in Pathology course, University of North Carolina

2000-2004	Course Director for Continuing Topics in Laboratory Medicine Series for Residents in Pathology, University of Pittsburgh Medical Center, Department of Pathology
1998-2004	Attending supervision and teaching of clinical residents and fellows in Pathology rotating on the Molecular Diagnostics Service. University of Pittsburgh Medical Center, Department of Pathology. ~200 contact hours per year.
2004	Lecture on "Emerging Infectious Diseases: West Nile Virus and SARS." Molecular Pathogenesis of Infectious Disease Course, MSI, University of Pittsburgh School of Medicine. 1 contact hour, ~150 first year medical students
2000-2004	Lectures on "Introduction to Virology," "Antiviral Therapy," and "Blood/transplant borne infections," Molecular Pathogenesis of Infectious Disease Course, 1 st year medical students, University of Pittsburgh School of Medicine. 5 contact hours, ~150 first year medical students per year
2001-2004	Problem Based Learning lab preceptor for Molecular Pathogenesis of Infectious Disease Course, 1 st year medical students, University of Pittsburgh School of Medicine. 12 contact hours, ~10 first year medical students per year. Received highest evaluation of all course preceptors in 2001.
2001-2003	Lecture on "Hepatitis Viruses: Acute and Chronic Hepatitis" and proctor for student journal club, Molecular Pathobiology Course (Graduate Level), University of Pittsburgh School of Medicine. 2 contact hours, 12 Graduate students
2001-2003	Faculty Mentor for MSI Journal Club, University of Pittsburgh School of Medicine. <i>4 contact hours, ~20 first year medical students per year.</i>
2000-2003	Lectures on "Hepatitis C: molecular testing in diagnosis and management" and "Clonal Analysis of Hematolymphoid Disorders," Didactic Lecture Series for Residents in Pathology, University of Pittsburgh School of Medicine. 2 contact hours, ~10 pathology residents per year
2001	Lectures on "Human Cytomegalovirus molecular biology" and "Animal herpesviruses as models." Advanced Topics in Herpesviruses Course (Graduate Level), University of Pittsburgh School of Medicine. 2 contact hours, 12 graduate students
2001	Problem Based Learning lab preceptor for Molecular and Human Genetics course, MSI, University of Pittsburgh School of Medicine. Covered Down Syndrome, Prader-Willi, Huntington Disease, Fragile X Disease, and Neurofibromatosis. 12 contact hours, 9 first year medical students
1999-2001	Lecture "Hepatitis Viruses," Viral Pathogenesis Course, Dept. of Infectious Diseases and Microbiology, Graduate School of Public Health, University of Pittsburgh. 2 contact hours, ~15 graduate students per year
1999	Course Director for Review Course in Molecular Diagnostics for Clinical Technologists, University of Pittsburgh School of Medicine, Division of Molecular Diagnostics.
1996	Markey Pathway Lecture Series, "EBV Vaccine strategies," Markey Graduate Students, Washington University School of Medicine, St. Louis, MO

1992-1996	996 Lecture on "Introduction to Clinical Medicine/ History and Physical Exam," to Occupational Therapy students, Washington University School of Medicine, St Louis, MO	
1991-1993	Course Director for Third Year Medical Student Lecture Series in Laboratory Medicine, Washington University School of Medicine, St. Louis, MO	
1988-1993	Lecture on "Blood Components and Transfusion Reactions," Third Year Medi Student Laboratory Medicine Lecture Series, Washington University School of Medicine	
1988-1990	Practical Lab Preceptor: "Blood and Urine Cultures," Second Year Medical Students, Washington University School of Medicine, St. Louis, MO	
RESEARCH	<u>I</u>	
Grant fundii	<u>ng</u>	
1U19HD077 0 NNGRI NC NEXUS: Co-investiga	9/05/13-8/31/2018 North Carolina Newborn Exome Sequencing in Universal Screening	
	87 (b) (c) PI) 12/05/11 – 11/30/15 North Carolina Clinical Genomic Evaluation by NextGen Exome Sequencing Investigator/ (c) (d) Effort	
NC Translati	nslational Science Award (CTSA) 2008-2113 ional and Clinical Sciences (TraCS) Institute NIH/NCRR \$7,352,734 Core Investigator/	
KG100355 Susan G. Ko Validating C' Co-Investiga	YP2D6 Genotype-guided Tamoxifen Therapy for a Multiracial U.S. Population	
UNC Lineber Co-sponsors	the Role of Genotype in Tamoxifen Therapy for Breast Cancer (PI: 1016) rger Comprehensive Cancer Center, University Cancer Research Fund s: LabCorp, Roche Molecular Diagnostics attor/ 1016 effort 2008-2010 UNC/UCRF)
UNC Investr	ments for the Future Grant (PI: ^{[5)(6)} Gap- <u>Bring</u> ing Genetics to Clinical Medicine	
	am in Translational Science (PI: 1016) at of a Translational Renal Genetics Program ector/ 1016 effort 2006-2008 UNC	
NCBC 2006-	-MRG-1117 (PI ^{(b) (6)}	

Development of Novel Diagnostic Testing for Primary Ciliary Dyskinesia Multidisciplinary Research Grant (MRG)/North Carolina Biotechnology Center (NCBC) Principal Investigator/ effort 2006-2008 NCBC

Collaboration Education and Test Translation Program (CETT)/

NIH Office of Rare Diseases (PI:

Development of clinical genetic testing for primary ciliary dyskinesia

Principle co-investigator 2006-2007 NIH ORD \$26,000

1U01DK060329

Multicenter Virahep-C Study of Viral Resistance to Antiviral Therapy of Chronic Hepatitis C

2001-2005 Consultant/ 3% effort

R01 Al51227-01 (PI: Randhawa)

Polyomavirus Infection After Kidney Transplantation

Co-Investigator/ 10% effort 2003-2005 NIH \$175,000

1 RO3 DK 60619-01 (PI: Shakil)

Outcome Of Hepatitis C Following Liver Transplantation

Consultant/ 3% effort \$200,000 2003-2005 NIH

Roche Study

The Utility of MecA Gene PCR Assays for Detection of MRSA Roche

Principle Investigator/ Effort 2003-2005

50795K (K08 Award)

Regulation of the EBV Lytic Switch Gene BZLF1

Principle Investigator /100% effort 1995-1998 NIH

Inventions

2001 Method for detecting BK virus and related compositions (Co-inventor), licensed

to Viracor laboratory, St. Louis, MO

PROFESSIONAL SERVICE

Membership In National Societies

American College of Medical Genetics (ACMG) Affiliate Specialist 2012-present Fellow, College of American Pathologists (FCAP) 2005-present American Society of Human Genetics (ASHG) 2004-present

Academy of Clinical Laboratory Physicians and Scientists (ACLPS) 1993-1998, 2006-present

American Association for the Advancement of Science (AAAS) 2000-2006 American Society of Microbiology (ASM) 1999-2005 Pan American Society for Virology (PASCV) 1998-2005 American Society for Virology (ASV) 1998-2005

Service on National Committees

2014-present	Clinical and Laboratory Standards Institute (CLSI) Consensus Committee
	on Molecular Methods, Advisor
2013-2014	Nominating Committee, Association of Molecular Pathology Solid Tumors Subdivision (elected office)
2013	College of American Pathologists House of Delegates member
2012- present	Chair, Biochemical and Molecular Genetics Resource Committee, College of American Pathologists/ American College of Medical Genetics
2012-present	Chair, Pharmacogenetics Workgroup, College of American Pathologists
2011-present	Member Molecular and Clinical Genetics Devices Panel of the US FDA Medical Devices Advisory Committee
2011-2013	Next Generation Sequencing Workgroup, College of American Pathologists
2009-2011	Chair, Training and Education Committee and Council Member, Association for Molecular Pathology (elected office)
2009-2010	Pharmacogenetics Workgroup, College of American Pathologists
2005-2010	Biochemical and Molecular Genetics Resource Committee, College of American Pathologists/ American College of Medical Genetics
2008	ABL Mutation Working Group, Association for Molecular Pathology Clinical Practice Committee
2007	Chair of the Program Committee and Council Member, Association for Molecular Pathology (elected office)
2006	Chair Elect of the Program Committee, Association for Molecular Pathology (elected office)
2004	Chair of Infectious Disease Subdivision and Council Member, Association for Molecular Pathology (elected office)
2003	Chair Elect of Infectious Disease Subdivision, Association for Molecular Pathology (elected office)
2000-2001	Infectious Disease Representative to the Training and Education Committee, Association for Molecular Pathology (elected office)
Consultancies	
2012-present	Consultant Advisory Panel, BlueCross BlueShield of North Carolina
2011-present	Member FDA Molecular and Clinical Genetics Devices Panel of the Medical Devices Advisory Committee
2010-2013	Consultant Laboratory Director, Gentris Corporation, Morrisville, NC
2006-2012	Advisory Board Member, Roche Molecular Diagnostics
2008-2010	Consultant to the FDA Molecular and Clinical Genetics Devices Panel of the Medical Devices Advisory Committee
2008-2010	Advisor, World Science Festival, New York City, NY http://www.worldsciencefestival.com

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College of American Pathologists Liaison to Model Genetics Test Report Workgroup, RAND Corporation/ CDC-funded
 Consultant Laboratory Director, ParagonDx, Morrisville, NC
 Consultant, Third Wave Technologies
 Ad Hoc consultant to McKenzie & Company, Gerson Lehrman Group,

and Easton Associates

Editorial Boards

2013 - present American Journal of Pathology

2010-present Expert Review of Molecular Diagnostics

2010-present Genetics in Medicine, Associate Editor of Molecular Genetics and

Pharmacogenetics

2009-2010 Genetics in Medicine, Associate Editor of Pharmacogenomics and

Personalized Medicine

2006-present Journal of Molecular Diagnostics

Other Service

2003-present Ad Hoc Molecular Laboratory Inspector, College of American Pathologists

1999-present Ad Hoc Reviewer for Transplantation, Journal of Virology, Journal of

Clinical Virology, Molecular Diagnosis, Bone Marrow Transplantation,

Molecular and Cellular Probes, Clinical & Diagnostic Laboratory

Immunology, Human Mutation, Clinica Chimica Acta, Clinical Chemistry,

Gene, and Laboratory Medicine

University of North Carolina

2004 – present Director Molecular Genetics, UNC Hospitals McLendon Clinical

Laboratories

2010 -present Department of Pathology and Laboratory Medicine Research Advisory

Committee

2013-present NC TraCS Institute/CTSA Translational Advancements Resource

Committee

BIBLIOGRAPHY

Refereed Primary Articles:

 Laurin LP, Lu M, Mottl AK, Blyth ER, Poulton CJ, Weck KE. Podocyte-associated gene mutation screening in a heterogeneous cohort of patients with sporadic focal segmental glomerulosclerosis. Nephrol Dial Transplant Feb 4, 2014 [epub ahead of print]. Article highlighted in Nature Reviews Nephrology.

- 2. Fan Z, Greenwood R, Felix ACG, et al. GCH1 heterozygous mutation identified by whole-exome sequencing as a treatable condition in a patient presenting with progressive spastic paraplegia. J Neurol Feb 8, 2014 [epub ahead of print].
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 (b)(4)
- 5. Jonas DE, Evans JP, Mcleod HL, Brode S, Lange LA, Young ML, Bryant Shilliday B, Martensen M, Swinton-Jenkins N, and Weck, KE. Impact of genotype-guided dosing on anticoagulation visits for adults starting warfarin: a randomized controlled trial. Pharmacogenomics 2013; 14(13): 1593–1603. PMID:24088130
- 6. Tarczy-Hornoch P, Amendola L, Aronson SJ, Garraway L, Gray S, Grundmeier RW, Hindorff LA, Jarvik G, Karavite D, Lebo M, Plon SE, Van Allen E, Weck KE, White PS, Yang Y. A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record. Genetics in Medicine 2013; 15:824-832. PMID:24071794
- 7. Mottl AK, Mei L, Fine CA, Weck KE. A novel TRPC6 mutation in a family with podocytopathy and clinical variability. BMC Nephrology 2013;14:104. PMID:23663351
- 8. Perera MA, Cavallari LH, Limdi NA, Gamazon ER, Konkashbaev A, Daneshjou R, et al. Genetic variants associated with warfarin dose in African American individuals: a genome wide association study. Lancet. 2013 Aug 31;382(9894):790-6. PMID: 23755828
- Scheuner MT, Hilborne L, Brown J, Lubin IM; members of the RAND Molecular Genetic Test Report Advisory Board. A report template for molecular genetic tests designed to improve communication between the clinician and laboratory. Genet Test Mol Biomarkers. 2012 Jul;16(7):761-9. PMID:22731646
- Hayes DN, Lucas AS, Tanvetyanon T, Krzyzanowska MK, Chung CH, Murphy B, Gilbert J, Mehra R, Moore D, Sheikh A, Hoskins JM, Hayward MC, Zhao N, Weck KE, Cohen RE, Cohen EE. Phase II efficacy and pharmacogenomic study of selumetinib (AZD6244; ARRY-142886) in iodine-131 refractory papillary thyroid carcinoma (IRPTC) with or without follicular elements. Clin Cancer Res. 2012 Apr 1;18(7):2056-65. PMID:22241789
- 11. Weck KE, Zehnbauer B, Datto M, Schrijver I. Molecular genetic testing for fragile X syndrome: laboratory performance on the College of American Pathologists proficiency surveys (2001-2009). Genet Med. 2012 Mar;14(3):306-12. PMID: 22241100
- 12. Pont-Kingdon G, Gedge F, Wooderchak-Donahue W, Schrijver I, Weck KE, Kant JA, Oglesbee D, Bayrak-Toydemir P, Lyon E. Design and analytical validation of clinical DNA sequencing assays. Arch Pathol Lab Med. 2012 Jan;136(1):41-6. PMID: 22208486
- 13. Lacbawan FL, Weck KE, Kant JA, Feldman GL, Schrijver I. Verification of performance specifications of a molecular test: cystic fibrosis carrier testing using the luminex liquid bead array. Arch Pathol Lab Med. 2012 Jan;136(1):14-9. PMID: 22208482
- 14. Davies JM, Trembath D, Deal AM, Funkhouser WK, Calvo BF, Finnegan T, Weck KE, Tepper JE, O'Neil BH. Phospho-ERK and AKT status, but not KRAS mutation status, are

- associated with outcomes in rectal cancer treated with chemoradiotherapy. Radiat Oncol. 2011;6:114. PMID: 21910869
- Irvin WJ Jr., Walko CM, Weck KE, Ibrahim JG, Chiu WK, Dees EC, et al. Multicenter study of genotype-guided tamoxifen dosing increases active metabolite exposure in women with reduced CYP2D6 metabolism. J Clin Oncol 2011;29:3232-9. PMID: 21768473
- Berg JS, Evans JP, Leigh MW, Omran H, Bizon C, Mane K, Knowles MR, Weck KE, and Zariwala MA. Next generation massively parallel sequencing of targeted exomes to identify genetic mutations in primary ciliary dyskinesia: Implications for application to clinical testing. Genet Med. 2011; 13: 218-229. PMID: 21270641
- 17. Limdi NA, Wadelius M, Cavallari L, et al. International Warfarin Pharmacogenetics Consortium. Warfarin pharmacogenetics: a single VKORC1 polymorphism is predictive of dose across 3 racial groups. Blood 2010 May 6;115(18):3827-34.
- Pratt VM, Zehnbauer B, Wilson JA, et al. Characterization of 107 Genomic DNA Reference Materials for CYP2D6, CYP2C19, CYP2C9, VKORC1, and UGT1A1: A GeT-RM and Association for Molecular Pathology Collaborative Project. J Mol Diagn 12: 835-846, 2010.
- 19. Kimani J, Buchman CA, Booker JK, Huang BY, Castillo M, Powell CM, Weck KE. Sensorineural hearing loss in a pediatric population: association of congenital cytomegalovirus infection with intracranial abnormalities. Arch Otolaryngol Head Neck Surg;136(10):999-1004, 2010.
- 20. Orsi FA, Annizzio JM, de Paula EV, Ozelo MC, Langley MR, Weck KE. VKORC1 V66M mutation in African Brazilian patients resistant to oral anticoagulant therapy. Thrombosis Research 126: e206–e210, 2010.
- 21. Barker CD, Bale S, Booker J, Buller A, Das S, Friemand K, Godwin AK, Grody WW, Highsmith E, Kant JA, Lyon E, Mao R, Monaghan KG, Payne DA, Pratt VM, Shrijver I, Shrimpton AE, Spector E, Telatar M, Toji L, Weck K, Zehnbauer B, and Kalman L. Development and Characterization of Reference Materials for *MTHFR*, *SERPINA1*, *RET*, *BRCA1*, and *BRCA2* Genetic Testing. J Mol Diagn 11(7): 553-561, 2009.
- 22. Langley MR, Booker JK, Evans JP, McLeod HL, Weck KE. Validation of Clinical Testing for Warfarin Sensitivity. Comparison of CYP2C9-VKORC1 Genotyping Assays and Warfarin-Dosing Algorithms. J Mol Diagn 11(3): 216 –225, 2009.
- 23. International Warfarin Pharmacogenetics Consortium. Estimation of the Warfarin Dose with Clinical and Pharmacogenetic Data. N Engl J Med 360:753-764, 2009.
- 24. Singh HK, Andreoni K, Madden V, True K, Detwiler R, Weck K and Nickeleit V. Presence of Urinary Haufen Accurately Predicts Polyomavirus Nephropathy. J Am Soc Nephrol 20:404-415, 2009.
- 25. Jones D, Kamel-Reid S, Bahler D, Dong H, Elenitoba-Johnson K, Press R, Quigley N, Rothberg P, Sabath D, Viswanatha D, Weck K and Zehnder J. Laboratory Practice Guidelines for Detecting and Reporting BCR-ABL Drug Resistance Mutations in Chronic Myelogenous Leukemia and Acute Lymphoblastic Leukemia. J Mol Diag 11(1):4-10, 2009.
- 26. Putcha GV, Otani IM, Khababa I, Booker J, Zariwala M, Weck K and Schrijver I. A Multicenter Study of the Frequency and Distribution of GJB2 and GJB6 Mutations in a Large North American Cohort. Genetics in Medicine 9(7):413-26, 2007.
- 27. Scanga L, Chaing S, Powell C, Aylsworth AS, Harrell LJ, Henshaw NG, Civalier CJ, Thorne LB, Weck K, Booker J, Gulley ML. Diagnosis of human congenital cytomegalovirus

- infection by amplification of viral DNA from dried blood spots on perinatal cards. J Mol Diagn 8(2):240-245, 2006.
- 28. Tarakanova VL, Suarez F, Tibbetts SA, Jacoby MA, Weck KE, Hess JL, Speck SH, Virgin HW 4th. Murine gammaherpesvirus 68 infection is associated with lymphoproliferative disease and lymphoma in BALB beta2 microglobulin-deficient mice. J Virol 79(23):14668-79, 2005. PMID:16282467
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